DISEASE:
Proximal spinal muscular atrophy type 4

NAME:	Proximal spinal muscular atrophy type 4
DESCRIPTION:	A rare, genetic proximal spinal muscular atrophy characterized by degeneration of alpha motor neurons in the anterior horns of the spinal cord and lower brain stem manifesting with adult onset, slowly progressive, mild proximal muscle weakness.
ORPHACODE:	83420
SYNONYMS:	SMA type 4 SMA type IV SMA-IV SMA4 Spinal muscular atrophy, adult form
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SMN2 SMN1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
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- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4 (Full sequencing)
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4 (SMN1 & SMN2 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- survival of motor neuron 1, telomeric
- survival of motor neuron 2, centromeric

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
